Educational Material Provided

Every infant born in the State of Tennessee is tested for rare, treatable disorders. The Newborn Screening Program provides the parents/guardian with an educational pamphlet to inform them about the testing and also addresses some of the most common questions asked about Newborn Screening. Signing below indicates that the facility where your baby was born provided you with the pamphlet.

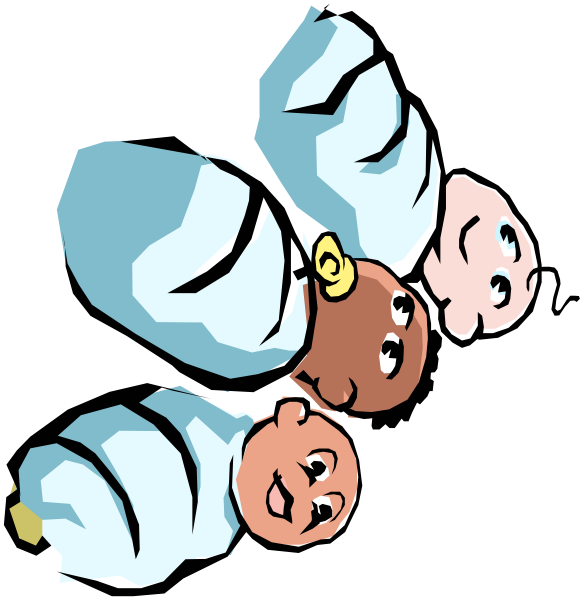
On _____/_____/_____, I _____ was provided with
Date Parent or Guardian

an educational pamphlet about Newborn Screening.

Witness:_____ Date:_____/_____/_____

Facility Staff:
Remove proof of documentation that educational material was provided and place in patient record.

THE METABOLIC/GENETIC
NEWBORN SCREENING PROGRAM
FOR THE STATE OF TENNESSEE



Your Baby and Newborn Screening

What is Metabolic Newborn Screening?

Testing for rare treatable disorders of body chemistry.

Who is Screened?

Your baby and every baby born in Tennessee.

When is it done?

Before you take your baby home from the hospital or between 24-48 hours after birth if your baby is not born in a hospital.

How is the testing done?

Before you take your baby home from the hospital, the nurse or laboratory technician will obtain a few drops of blood from your baby's heel. The blood is absorbed onto a special filter paper, and sent to the state laboratory for testing. This testing will determine if your baby may have any of these disorders. **It is important to note that these are screening tests, not diagnostic tests. More testing will need to be done if the screening test is abnormal.**

Why is it done?

Babies with these disorders usually appear completely normal at birth. However, without treatment, they can become very sick and/or mentally retarded. These bad effects can be prevented or reduced with treatment if the disorders are detected early.

Where is it done?

The blood sample is taken in the hospital or birthing center. The testing is done at the State Newborn Screening Laboratory in Nashville.

Amino Acid Disorders – Amino acid disorders are a group of conditions in which there is a problem with breaking down certain components of food called amino acids. These disorders are caused by a specific defect in one of the many enzymes that perform these tasks. The specific amino acid can build up in the blood and other organs, including the brain. This amino acid and any of its metabolites can cause serious health problems such as mental retardation, damage to vital organs, seizures or coma. The effects of the disorder will vary, and depend on the age at which symptoms occur and the specific amino acid(s) elevated. Treatments vary and may include special dietary intervention, replacement medications, acute illness protocols, and metabolic genetic and nutritional monitoring.

Organic Acid Disorders – Organic acid disorders are a group of conditions in which there is a problem with breaking down protein and amino acids in foods. This is due to a specific defect in one of the enzymes that breaks down these substances. These organic acids can build up in blood and urine, and can lead to problems such as low blood sugar, failure to thrive, developmental delays, infections and in rare occasions coma and death. The effects of the disorder will depend on the age at which symptoms occur. Delay in the recognition and treatment may have serious consequences. Treatment may include special dietary intervention, replacement medications, acute illness protocols, and metabolic genetic and nutritional monitoring.

Fatty Acid Oxidation Disorders - Fatty Acid Oxidation Disorders are a group of conditions that affect the breakdown of certain fats called fatty acids. A defect in a specific enzyme leads to a build up of fatty acids in the body. When a baby with one of these conditions "fasts" (goes for a long period of time without eating), problems can happen. This occurs because the baby cannot use the energy stored in the fats of the body. This kind of metabolic crisis can sometimes lead to seizures, failure to breathe, cardiac arrest, and death. It is extremely important to identify a child with this disease so that crisis can be prevented. Treatment may include avoiding fasting, replacement medications, monitoring the diet for specific metabolic nutritional requirements and blood levels of certain metabolites.

But my baby seems very healthy. Are the tests still necessary?

YES! Most babies with these disorders show no obvious signs of disease at birth. There is an "invisible" problem in one of the many chemicals that are produced in the baby's body. The special screening tests detect these chemical changes before problems develop. By testing every baby shortly after birth, we can be sure that a baby who has one of these disorders will be identified and started on treatment early.

If my baby has one of these disorders, can it be cured?

No, not really. It cannot be cured, just as eye color or height can't be permanently changed. However, the serious effects of the disorder can be lessened and often completely prevented if a special diet or other medical treatment is started early.

Will I receive a report of the test results?

Your doctor or health department will be informed of the results. Generally parents are notified only if repeat testing is needed. You can ask about the result when you take your baby to the doctor for a regular checkup.

What happens if one of the tests' results is "abnormal"?

If any of the tests are abnormal, showing a possible disorder, the follow-up program will contact your health department or baby's doctor and a specialist from the metabolic/genetic network immediately to request another blood sample. You will be asked to bring your baby in for a retest as soon as possible. Prompt action is very important. For confirmation and treatment, your baby will be referred to a specialist. You should make sure the hospital where your baby is born has your correct name, address and phone number. If you child should need to be retested, your baby's doctor will know where to reach you. Remember, time is very important.

If a retest is necessary, does that mean that my baby is sick?

Not necessarily. Retesting may be required for a number of reasons such as the first test was improperly collected, the baby received a blood transfusion, the specimen was collected when your baby was less than 24 hours of age or it could indicate a possible disorder. While taking your baby in for retesting can be scary, it is important that every baby has a thorough screening for all the disorders. Only on a very rare occasion will the doctor insist on treating the baby immediately while waiting for the results of the second test.

If my baby has a disorder, will my future children have it also?

Possibly. Families who have a child with one of these disorders should obtain information about their future risks from trained professionals with the Tennessee Genetics Network. See the list in this pamphlet for the center in your region.

What is Newborn Hearing Screening?

Hearing screening is not a blood test. Babies can have their hearing checked soon after birth. The test is very safe and does not hurt. Your baby may "pass" the hearing test or may need to be "referred" for further testing. Half of all babies identified with a hearing loss do not have a known cause for hearing loss. Babies identified with a hearing loss will be encouraged to be evaluated at a genetic center. Only 10 percent of babies with a hearing loss are born to parents who have a hearing loss. Significant hearing loss in both ears is present in 1-3 per 1000 newborns in the well baby nursery population, and 2-4 per 1000 in the intensive care unit population. For more information, call 615-741-8530 or 615-262-6160.

Maple Syrup Urine Disease (MSUD) – Occurs in about one out of every 230,000 babies born. MSUD occurs when a baby's body is not able to properly break down several amino acids, which are found in the protein of food. The build up of these amino acids in the body causes the baby's urine to smell like maple syrup. It can cause mental retardation, seizures or death if not treated. When detected early, the baby is put on a special low protein diet to help avoid the severe effects of the disease.

Medium-Chain Acyl CoA dehydrogenase (de-hi-dro-jen-as) (MCAD) Deficiency – Occurs in about one out of every 12,000 babies born. This disorder is caused by the lack of an enzyme that breaks down fat stored in the body. When a baby "fasts" (goes for a long period of time without eating), problems can happen because the baby cannot use stored fat properly. This kind of metabolic crisis can sometimes lead to seizures, failure to breathe, cardiac arrest, and death. Treatment is effective and focuses on avoiding fasting for long periods.

Biotinidase Deficiency (bi-oh-tin-I-das) – Occurs in about one out of every 61,000 babies born. This disorder is caused by the lack of an enzyme in the baby's body called biotinidase. Babies with biotinidase deficiency can have seizures, feeding difficulties, illness soon after birth, low muscle tone, skin rash or infection, developmental delays and hearing loss. Problems with the disorder can be prevented with biotin treatment.

Hospital Staff:

Review the following information with parents if the specimen was collected <24 hours of age and baby is being discharged home.

To be sure the screening results are accurate, babies who go home and have had a newborn screening specimen collected prior to 24 hours of age must be rescreened. Your baby's specimen was collected when he or she was less than 24 hours old and will need to be repeated within 24-48 hours. Take your baby to your pediatrician or local health department to have the specimen recollected.

Questions?

For more information about newborn screening: contact your baby's doctor, local health department or the Tennessee Newborn Screening Program at (615) 262-6304. Visit our website at: www2.state.tn.us/health/mch/genetics.htm

What are the disorders? Your child will be tested for the most common and/or most severe forms of the following disorders:

PKU or Phenylketonuria (Fen-il-ke-to-nu-ree-ah) – Occurs in about one out of every 14,000 babies born. Babies with PKU do not have a body chemical (enzyme), which is needed to break down part of the protein in food. This protein part, called phenylalanine, can build up in the brain and cause mental retardation. A special diet low in phenylalanine can prevent mental retardation and other effects of PKU.

Galactosemia (ga-LAK-toe-see-mi-ah) – Occurs in about one out of every 53,000 babies born. Babies with galactosemia do not have an enzyme needed to break down galactose, a kind of sugar found in milk. Babies with galactosemia can become very sick after a few days of normal feeding.

If not treated, they often develop eye problems and mental retardation. Babies with untreated galactosemia also have a high risk for infections, which can cause infant death. Galactosemia can be treated by putting the baby on a special galactose free diet. Some infants may have a milder form of galactosemia, which may require treatment for 6-12 months.

Congenital Hypothyroidism (con-gen-i-tal hi-po-thi-royd-ism)– Occurs in about one out of every 3,000 babies born. Hypothyroidism occurs when the body does not make enough thyroid hormone. Thyroid hormone is needed for brain and body growth. Babies with hypothyroidism may not grow well and be mentally retarded. If hypothyroidism is detected early and the baby is given medicine, normal growth and development can take place.

Hemoglobinopathies (he-mo-glo-bi-nop-a thes)– Hemoglobin is the part of the red blood cells which makes them look red and carries oxygen to the body. Hemoglobinopathies are diseases that affect the kind or the amount of hemoglobin a person has in the red blood cells. Some hemoglobinopathies can cause anemia.

Sickle cell disease is the most common hemoglobinopathy. It causes misshaped (sickle-shaped) red blood cells. These sickle red blood cells can clog blood vessels so that parts of the body do not get enough oxygen. Good medical care, parent education and antibiotics can lessen the life threatening complications of hemoglobinopathies.

Hemoglobinopathies can occur in all racial groups. The most common form, sickle cell disease, occurs more often in African Americans with the frequency of about one out of every 500 babies born. People of Hispanic, Asian, Arabic or Mediterranean backgrounds are also more likely to have a hemoglobinopathy.

CAH or Congenital Adrenal Hyperplasia (con-gen-I-tal ad-re-nal hi-per-play-see-ah) – Occurs in about one out of every 19,000 babies born. CAH occurs when the baby cannot make certain hormones. This results in abnormal hormone levels, which can cause infants to become very sick and if not treated, could even lead to death. Baby girls with this disorder may also be assigned the wrong gender at birth. CAH can be treated with medication.

Homocystinuria (ho-mo-sis-tin-u-re-ah) – Occurs in about one out of every 340,000 babies born. This disorder occurs when the body cannot process a chemical called homocystine, which builds up in the blood causing mental retardation, eye problems, and failure of the body to grow and gain weight. Blood clots can develop and potentially lead to life threatening problems. When detected early, babies with Homocystinuria are put on a special diet to help avoid these problems.

Genetics Network

Genetic/Metabolic Centers

University of Tennessee, Memphis (901) 448-6595
Vanderbilt University, Nashville (615) 322-7601
University of Tennessee, Knoxville (865) 544-9030

Satellite Genetic Centers

T.C. Thompson Children's, Chattanooga (423) 778-6112
East Tennessee State Univ., Johnson City (423) 439-8541

Hematology/Sickle Cell Centers

Pediatric Hematology Center, Memphis (901) 495-5670
Meharry Sickle Cell Center, Nashville (615) 327-6763
University of Tennessee, Knoxville (865) 544-9030
T.C. Thompson Children's, Chattanooga (423) 778-7289

Pediatric Endocrinologists

University of Tennessee, Memphis (901) 572-5096
Endocrine Clinic, Memphis (901) 763-3636
Jackson Pediatric Center, Jackson (731) 664-9928
Vanderbilt University, Nashville (615) 322-7427
East Tenn. Children's Hospital, Knoxville (865) 971-7400
T.C. Thompson Children's, Chattanooga (423) 778-6405
East Tennessee State Univ., Johnson City (423) 439-8541